# Finding the gaps

Access to Australian Government funding support for children with undiagnosed or rare genetic conditions.

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### Research team

Fleur Smith, Nossal Institute for Global Health, The University of Melbourne

Maxwell Thanh Le, Master of Public Health student, The University of Melbourne

Heather Renton, Syndromes Without A Name (SWAN) Australia

Dr Manjula Marella, Nossal Institute for Global Health, The University of Melbourne

### Acknowledgements

This study received funding from the Melbourne Disability Institute's Community-Based Research Scheme, with in-kind contributions from the Nossal Institute.

Parts of this study were undertaken by Maxwell Thanh Le as a research project for the Master of Public Health.

We thank all the members of SWAN's Consumer Advisory Group (Ros Melling, Sara Crawford, Maya Pinn, Samantha Steele, Jenny Downing, Julie Edwards, Amanda Lennestaal, Joel Taggart) for their valuable time and contributions to the design and testing of the data collection tools for this study. We would also like to thank Ant Howard for her inputs and support throughout the project.

Special thanks to all the survey and interview participants for generously sharing their experiences and perspectives.

## Abbreviations

| | |
|---|---|
| **ADL** | Activities of daily living |
| **AT** | Assistive Technology |
| **CALD** | Culturally and linguistically diverse |
| **CEO** | Chief Executive Officer |
| **CAG** | Consumer advisory group |
| **DSP** | Disability Support Payment |
| **ECEI** | Early Childhood Early Intervention |
| **ESS** | Education Support Supplement |
| **LAC** | Local Area Coordinators |
| **NDIA** | National Disability Insurance Agency |
| **NDIS** | National Disability Insurance Scheme |
| **PLS** | Plain language statement |
| **SPSS** | Statistical Package for Social Sciences |
| **SWAN** | Syndromes Without A Name |

# Executive summary

Rare genetic conditions affect approximately 1 in 2000 Australians, including 400,000 children [1]. Many people with a rare genetic condition wait years for a confirmed diagnosis, or never receive a diagnosis. Diagnostic challenges impact understanding of their condition and accessing the right supports.

People living with undiagnosed or rare genetic conditions frequently require a range of supports to enable their health, wellbeing, inclusion and participation in all areas of daily life. Accessing these supports can create significant financial pressure on individuals and their families from direct out of pocket service costs and indirect costs such as transport, equipment, medications and loss of household income. Many day-to-day supports are provided by family members, often at the expense of income generation. These financial pressures place individuals with undiagnosed or rare conditions at risk of missing out on the supports they need to live a good life, and places households under physical and psychological stress.

Families caring for a child with disability, including those with undiagnosed or rare genetic conditions, can apply for a range of Australian government funded financial support schemes [2]. These schemes are available to help families with the added costs associated with caring for and supporting a person with a disability. However, Syndromes Without A Name (SWAN) Australia, the only national organisation providing information, support, connection and advocacy for families caring for children with a undiagnosed or rare genetic condition, had heard anecdotal reports suggesting that those without a diagnosis, or with a rare diagnosis that is not well-known, face barriers to accessing these schemes.

This study aimed to understand the experiences of accessing Australian government funding support schemes for families of children with undiagnosed or rare genetic conditions, how funding is used, to identify current barriers and impacts of lack of access to funding, and opportunities to improve access.

## Summary of methods

This study used a mixed methods study design incorporating both a quantitative survey of SWAN members, supplemented by a small number of qualitative interviews with parents or caregivers of children with undiagnosed or rare genetic conditions.

## Key findings

### Socio-demographics of respondents

The survey received a total of 129 responses, representing a total of 158 children, as some respondents had more than one child with an undiagnosed or rare condition (17%). Most respondents were mothers (96%) from metropolitan areas (63%). Responses were received from all states and territories except the Northern Territory. Out of the 158 children, approximately two thirds (65%) had a confirmed diagnosis, while one third had not yet received a diagnosis.

Eight interviews were conducted with parents of children with undiagnosed or rare genetic conditions, representing ten children in total. All but one respondent were female caregivers, six of the children had received a diagnosis.

### Support needs

Respondents described that the supports needed for their child and family as a result of their child's condition included support workers to assist with day to day caring and accessing activities; therapeutic supports; assistive technology; transport; home modifications; support for education; health and medical supports.

### Access to government funding schemes

Almost all participants had applied for and were receiving funding from at least one of the available funding schemes. The Carer Allowance and the National Disability Insurance Scheme (NDIS) were the most frequently applied for and accessed. The Pharmaceutical Allowance and Taxi Subsidy Scheme the least frequently applied for.

For some schemes, access was similar between those with and without a diagnosis (NDIS, pharmaceutical allowance, education support supplement), while for other schemes access was higher among those with a diagnosis compared to those without a diagnosis (Carer Payment, Carer Adjustment Payment, and Companion Card).

Of those who had applied for funding, approximately half reported that the funding received was insufficient to cover their child's support needs.

### Barriers to funding access

The most common barriers to accessing funding identified were: 1) lack of knowledge of available funding schemes; 2) difficulty demonstrating eligibility due to lack of diagnosis or having a rare diagnosis; and 3) systemic issues in the administration of funding schemes including complex application processes and lack of knowledge or understanding by staff of the needs of children with undiagnosed or rare conditions.

Participants reported a need for clearer, more timely provision of information about the funding schemes available. This information tended to depend on the knowledge of individual treating health professionals, who were often the main point of contact in the early stages of families' journeys.

Many respondents perceive that access to funding is more difficult without a diagnosis, or with a rare diagnosis that funding bodies are not familiar with, even if support needs are similar to children with more well-known conditions. This was raised particularly as an issue when accessing NDIS funding. Comments of needing to "jump through hoops" to explain their child's needs or that their child "doesn't fit in any of the boxes" due to not have a diagnosis were common.

Application processes were described as confusing, complex and arduous, even for those with high levels of literacy and English language proficiency. Obtaining reports and supporting documentation to support applications was time consuming and, often, costly. Access was further hampered by applications going missing, long waiting times for outcomes, conflicting information, and lack of knowledge by those deciding funding outcomes.

### Enablers of funding access

Factors that that assisted or enabled access to funding could be broadly grouped into 1) support from health professionals; 2) strategies implemented by the family; 3) funding scheme factors; and 4) seeking additional support.

Where treating medical and allied health professionals had good knowledge of the NDIS or other schemes and the documentation required, respondents reported that funding access was easier and more successful. Having the means to pay for additional assessments or reports, giving detailed carer statements, speaking to other parents, searching the internet and social media, and appealing decisions, were all reported as enabling factors that supported access to funding. Some participants also reported having access to an NDIS support co-ordinator or advocacy organisation was also helpful.

### Use and positive impact of funding

Funding received through government schemes was mostly reported to be used for therapy services to support children's skills and independence; assistive technology and equipment; support workers to assist with self-care, social or recreational activities; and respite or caregiving support. Smaller numbers of

participants reported using funding to help cover transport or health care costs, education related supports, or for home modifications.

Having access to funding was described as “life changing” by some participants, contributing to significant improvements in their child’s functioning and wellbeing. Funding also reduced the financial burden on families and the need to fundraise through other avenues. Access to funding for the key support needs allowed families to prioritise their own personal funds for things that can’t be funded through government funding but that were also considered important for their child’s or family’s wellbeing, such as holidays or other recreational activities

### Unmet support needs

Barriers to funding results in significant impacts for both children and families, including delayed or lack of access to needed supports, and financial stress leading to families using personal savings, borrowing money from families or friends, taking out loans, seeking funding from charities, and refinancing mortgages to cover costs.

Many households identified not having sufficient funding to access respite and caregiving support, and support for their child’s social and community participation as the greatest unmet needs. These unmet needs had subsequent impacts on child participation in activities with peers, household financial stress, caregiver wellbeing and family relationships.

## Recommendations

The following recommendations are made to address barriers identified in this study and improve access to funding and support for children with undiagnosed or rare genetic conditions, and their families.

- Ensure clear and accessible information that increases parent’s/caregiver’s awareness of what funding and support is available to them from when a child’s is first identified as not meeting their milestones or has health concerns, even in the absence of a diagnosis.
- Promote information about funded supports, health and disability information including support groups to frontline health professionals (e.g. GPs, paediatricians, Maternal & Child Health Nurses, allied health professionals) who may be first point of contact.
- Implement support mechanisms for families to navigate funding application processes, including for appealing decisions. This could include developing peer support networks for navigating application processes, parent educations sessions specific to applying for funding for children with rare and undiagnosed genetic conditions, or an online resource repository to share templates or tips of how to complete application forms.
- Build capacity of agencies and workers responsible for assessing funding applications and determining eligibility for funding support to better understand the support needs and complexities for children with undiagnosed and rare genetic conditions. This could include access to a Rare Disease System navigator or focal point within the NDIA or Services Australia, who could support undiagnosed and rare disease families as well as government agencies and their staff.
- Funding eligibility should be determined on function and support needs, not diagnosis, including removal of NDIS defined disability lists.
- Provision made for interim funding while waiting for a diagnosis to reduce the uncertainty for these families, who are already experiencing the uncertainty of a diagnosis that is either unknown or rare with little information on their child’s prognosis.

# 1. Background

Rare diseases (or rare conditions) are those which affect less than five in 10,000 people [3]. In Australia, rare diseases have a prevalence of 1 in every 2000 people and are estimated to affect approximately 5-8% of the population [1, 4]. This is equivalent to approximately two million Australians living with a rare condition including 400,000 children [1, 5]. Although a disease can be rare, rare diseases collectively are relatively common with more than 7000 different types of rare conditions known and new conditions continuing to be discovered with developments in technology [6, 7].

Although undiagnosed or rare genetic conditions may result in different signs and symptoms, individuals with a rare condition and their families experience common challenges [3, 8, 9]. Rare diseases generally have a childhood onset and persist throughout adulthood causing long-term health impacts, and in some cases can be life limiting [1, 8]. Many individuals with a rare condition also live with intellectual or physical disability, or both, with significant implications to their functioning and everyday life[6, 10]. People with rare conditions and their family may also experience psychological, emotional, financial, physical, and psychosocial impacts as a consequence of additional health needs and functioning difficulties [11-14].

### Diagnostic challenges

People living with a rare disease frequently face challenges in receiving a timely and accurate diagnosis due to the lack of knowledge about rare diseases by health professionals, and limited access to genetic testing [15, 16]. On average, individuals with rare conditions and their families wait 5-9 years for a diagnosis, with 40% likely to receive at least two misdiagnoses before receiving a confirmed final diagnosis[17, 18]. An incorrect or delayed diagnosis can lead an individual to undergo unnecessary treatment and cause delays in receiving appropriate treatment and management [19-21]. Once diagnosed, many individuals and their families report continuing to feel isolated and under supported because of the lack of information known about the condition, limited available treatments or opportunities to participate in clinical trials or other research projects for their rare disease [6, 21, 22].

### Support needs and associated costs

People with rare genetic conditions may require specialist health services and the need for a wide range of other support services to enable optimal wellbeing, functioning and participation in everyday activities.

Health related needs can include frequent visits to medical specialists, allied health professionals, and the need for specialised equipment [15, 16]. These supports frequently incur out-of-pocket expenses, as well as loss of caregiver income and travel costs to attend appointments as well as the high cost of medications, many not covered by the Pharmaceutical Benefits Scheme. In addition, non-health related costs for individuals with an undiagnosed or rare genetic condition and their families may include respite care, child care, transport, home modifications, additional heating, food, communication support and cleaning expenses [23].

Meeting these costs can lead to financial hardship for families of children with an undiagnosed and rare conditions [24], and can be at increased stress and risk of family breakdown [3, 10].

### National Strategic Action Plan for Rare Diseases

In 2020, Australia launched the National Strategic Action Plan for Rare Diseases (hereafter 'the Action Plan') to equitably support and address rare diseases nationally [18]. The Action Plan included three pillars to drive progress for the rare disease population including 1) awareness and education, 2) care and support and 3) research and data. However, since the launch of the Action Plan only minimal efforts to support individuals with an undiagnosed or rare condition have occurred [1]. Families caring for children with an undiagnosed or rare disease have relied on self-research via the internet and peer support groups to find relevant information and support [6]. Many genetic or rare disease peer support groups, including SWAN, receive very little funding and are at risk of closure due to the heavy demands on them without the

resources to support them. Previous studies have called for further action to provide a coordinated response that can assist families with navigating the health system for a diagnosis and access to support services including financial support [3, 8, 12, 17]. Individuals with an undiagnosed or rare genetic condition still lack inclusive and appropriate services which meet their needs [11].

### Government funding

In Australia, individuals caring for a child with disability, including those with undiagnosed or rare genetic condition, can apply for a range of Australian government funded financial support schemes [2]. These schemes are available to help families with the added costs associated with caring for a person with a disability [2]. The available funding schemes include the Carers Allowance, Carer Payment, Carer Supplement, Carer Adjustment, Disability Support Pension (DSP), the National Disability Insurance Scheme (NDIS) and others (Table 1).

However, eligibility for these funded support options are often reliant on a known diagnosis result or medical records from a health professional to confirm the care needs of an individual (Table 1) [2]. This is an issue for many individuals with rare genetic conditions who undergo an extended process and wait time for a diagnosis, have an incorrect diagnosis or are undiagnosed [18]. For individuals with a diagnosis of a rare condition, access to funding supports can be equally challenging due to a lack of awareness and a lack of evidence about the condition and associated support needs [16]. Barriers in accessing these funds are likely to impact individuals with an undiagnosed or rare genetic condition, their families and carers from accessing needed support services [25].

**Table 1: Government funding schemes and their eligibility criteria[1]**

| Funding support options | Description | Eligibility criteria |
|---|---|---|
| **Carer Allowance** | This allowance provides an income supplement for carers or parents who provide daily care for an adult or dependent child or children. You may be able to get this payment if you care for someone with disability or a medical condition, or a frail older person, and if you also work or study. | • care for someone whose care needs score is high enough on the adult or child assessment tools<br>• care for someone who'll have these needs for at least 12 months<br>• meet an income test |
| **Carer Payment** | The Care Payment is a fortnightly payment which provides income support for individuals who cannot work because they care for someone with a severe disability, illness, medical condition or frail and old.<br>You may be able to get this payment if you provide constant care in a private home for someone with a severe disability or severe medical condition or who is frail and elderly, and if you aren't away from them for more than 25 hours a week to work. | • be an Australian resident<br>• care for someone who is an Australian resident<br>• care for 1 or more people who have care need scores high enough on the assessment tools used for an adult or child<br>• care for someone who'll have these needs for at least 6 months<br>• be under the pension income and assets test limits. |

[1] Except for NDIS, the data for descriptions in this table are from *Victoria – Family and carer support,* by Disability Gateway Department of Social Services, 2022 (https://www.disabilitygateway.gov.au/income-finance/family-carer-support/vic). Copyright 2022 by Department of Social Services. The data for Eligibility are from *Getting a Payment,* by Services Australia Australian Government, 2021 (https://www.servicesaustralia.gov.au/getting-payment-if-youre-carer?context=60097). Descriptions for NDIS are from https://www.ndis.gov.au/ [Accessed 4 March 2024].

| Funding support options | Description | Eligibility criteria |
|---|---|---|
| **Carer Adjustment Payment** | A one-off payment to assist families following a catastrophic event where a child younger than 7 is diagnosed with a severe disability or severe medical condition. | • you must get Carer Allowance for the child<br>• you must have a very strong need for financial help<br>• the child must need this care for at least 2 months<br>• you and your partner can't be getting Carer Payment or be able to get another income support payment from us. |
| **Carer Supplement** | This supplement is a yearly payment to help with the cost of caring for a person paid in July each year. You might be able to get this payment if you get a Carer Payment or a Carer Allowance. | You can get Carer Supplement if you get any of these payments:<br>• Carer Allowance<br>• Carer Payment<br>• Department of Veterans' Affairs Partner Service Pension and Carer Allowance<br>• Department of Veterans' Affairs Carer Service Pension.<br>You must get one of these payments for 1 July to be eligible. |
| **Disability Support Pension (DSP)** | Financial help if you have a permanent physical, intellectual or psychiatric condition that stops you from working. | To get DSP, you need to meet both:<br>• non-medical rules e.g. age, residence status, and income and assets.<br>• medical rules. |
| **National Disability Insurance Scheme (NDIS)** | The NDIS provides funding to eligible people with disability to gain more time with family and friends, greater independence, access to new skills, jobs, or volunteering in their community, and an improved quality of life. | To be eligible for the NDIS you must:<br>• Be aged under 65 years<br>• Be an Australian citizen, or hold a Permanent Visa or Protected Special Category Visa<br>• Live in Australia<br>• Have a disability caused by a permanent intellectual, cognitive, neurological, sensory, physical or psychosocial impairment<br>• Need disability-specific supports to complete daily life activities OR need supports to reduce future need for supports |
| **We Care Card (Victorian Carer Card)** | Provides a range of discounts and benefits from businesses, local government and community organisations for eligible carers. Entitles the holder to discounted travel on public transport. | To be eligible for the We Care Card you must be a resident of Victoria and either:<br>• the primary carer of a person with a disability, severe medical condition or mental illness, or someone who is frail aged or in need of palliative care<br>• a foster, kinship or respite carer |

## Study rationale

Currently a lack of information exists on the experiences of parents and others who care for children with an undiagnosed or rare genetic condition in accessing financial support from Services Australia and the National Disability Insurance Agency (NDIA) in Australia [2].

Few studies have examined the experiences of families accessing Australian government financial support for their child with an undiagnosed or rare condition. This study aimed to better understand what it is like for families of children with undiagnosed and rare genetic conditions to access government funding schemes, and how this may impact on their children and families' life.

Syndromes Without A Name (SWAN) Australia is the only national organisation providing information, support, connection and advocacy for families caring for children with a undiagnosed or rare genetic condition[26]. SWAN had received several reports from their members who have experienced significant difficulties in accessing or maintaining suitable government funded supports to meet their child's support needs. The existing funding schemes are hypothesised to not be accommodating for children with an undiagnosed or rare genetic condition and resulting in inequitable access to funded support by SWAN families. In contrast, individuals with more commonly recognised genetic conditions such as Down Syndrome or Fragile X are perceived to experience less difficulties in applying and accessing government financial support. SWAN was interested in understanding the challenges experienced by families caring for a child with an undiagnosed or rare genetic condition in accessing Australian government funding programs and to identify opportunities to provide better access to needed supports for SWAN children and their families.

## 2. Study aims

This mixed method study aimed to understand the experiences of accessing Australian government funding support schemes for families of children with undiagnosed and rare genetic conditions and identify current barriers and impacts of lack of access to funding.

### Research questions

- What is the experience for families of children with undiagnosed and rare genetic conditions in applying for and receiving Australian government funding support schemes?
- What are the barriers and facilitators to accessing government funding support schemes for this cohort?
- How does access to funding, or lack of access to funding, impact on children with undiagnosed and rare genetic conditions and their families?

## 3. Methods

This study used a mixed methods study design incorporating both a quantitative survey and qualitative interviews with parents or caregivers of children with undiagnosed and rare genetic conditions.

### 3.1. Data collection and participant recruitment

#### 3.1.1. Quantitative survey

An online survey was conducted using the platform Qualtrics. The survey included a plain language statement (PLS) with explanation of the purpose, benefits, risks and objectives of the study. Questions were in three sections 1) Respondent demographics; 2) Access to funding schemes; and 3) Understanding the barriers, enablers, and impact of access to funding. Respondents with more than one child with an undiagnosed or rare condition could repeat the survey questions under Section 2 to answer for each specific child. The survey questions were developed by the research team with input from SWAN's Consumer Advisory Group (CAG) who also tested the tool for overall acceptability and usability.

Survey data was collected for a three-week period from late July to mid-August 2022. Survey respondents were any parent, guardian or carer living in Australia who cared for a child or children with an undiagnosed or rare genetic condition.

The survey link was distributed via email by SWAN to their member database and promoted via their newsletter and social media. The link was also able to be forwarded by members to parents of children with undiagnosed and rare conditions who were not SWAN members. Informed consent was collected from all survey respondents at the start of the survey through a short PLS where respondents indicated their consent to proceed to survey questions.

#### 3.1.2. Qualitative interviews

Qualitative data was collected via semi-structured interviews with parents of children with undiagnosed and rare genetic conditions. These interviews were conducted by phone or video call (Zoom, Microsoft Teams) depending on the participants preference. Interviews were used to explore factors covered in the quantitative survey in more depth to understand the experiences of accessing government funding supports, the challenges, any facilitators, and the impacts for children and families, and potential systems improvements. A question guide was developed by the research team with input from the CAG. Interviews were recorded and transcribed.

Participants for the interviews were recruited via the survey. At the end of the survey, respondents were asked for their interest to participate in an in-depth qualitative interview and asked to provide their contact details if interested. Potential participants were purposively selected from those who expressed interest to

ensure representation from different states, and to include parents/caregivers of children of varying ages and numbers of children, and those with and without confirmed genetic diagnoses. These people were contacted and provided with a PLS about the interviews and provided consent prior to being interviewed. A total of eight interviews were conducted.

## 3.2. Data analysis

### 3.2.1. Quantitative survey

Data was exported from Qualtrics (https://www.qualtrics.com/) to Statistical Package for Social Sciences (SPSS) version 27 (SPSS, Inc., Chicago, Illinois). Prior to analysis, survey data was cleaned. Any incomplete surveys were not analysed. Survey data was de-identified and respondents were assigned a unique identification number for their household and each of their children. This ensured confidentiality and protected the small sample from being re-identified. Quantitative data was analysed using SPSS to generate descriptive statistics. Responses obtained from open-ended survey questions were analysed thematically. Initial findings were presented to SWAN's CEO for feedback and discussion.

### 3.2.2. Qualitative interviews

Data from the interviews was analysed using Nvivo software (release 1.7.1). Data was coded by two members of the research team and analysed inductively and deductively. The objectives of the research provided initial structure for thematic analysis, with emerging themes and subthemes added and data coded accordingly and triangulated with quantitative data.

## 3.3. Ethics

Ethics approval was obtained from the University of Melbourne Human Research Ethics Committee prior to the commencement of data collection.

# 4. Findings

## 4.1. Demographics

### 4.1.1. Survey respondents

The survey received a total of 129 responses, representing a total of 158 children. Table 2 shows the demographics of respondents caring for children with an undiagnosed or rare condition. The majority of respondents were mothers (n=124, 96.1%) with one SWAN child (n=107, 82.9%) from a metropolitan city across Australia (n=81, 62.8%). Responses were received from all states and territories across Australia except the Northern Territory. 22 (17%) households in the study had more than one SWAN child, with a maximum of four SWAN children in one household. 21 (16%) respondents identified as having disability, genetic or mental health condition themselves, with 42.9% (n=9) of these parents and guardians indicating their condition sometimes has an impact on their ability to apply for, manage or maintain government funded support for their children.

The demographics of respondents' children with an undiagnosed or rare condition are shown in Table 3. Children's age included 12 (1.9%) that were under 12 months, 40 (25.3%) aged 1 to 5 years, 74 (46.8%) aged 6-12 years, 28 (17.7%) aged 13 – 17, and 13 (8.2%) that were 18 years or older. Out of the 158 children, 102 children (64.6%) have a confirmed genetic diagnosis whilst 56 children (35.4%) have not yet been diagnosed.

**Table 2: Demographics of survey respondents**

| Demographics of respondents (n = 129) | N (%) |
|---|---|
| **Gender** | |
| Female | 124 (96.1) |
| Male | 5 (3.9) |
| **Aboriginal Australian** | |
| Aboriginal | 1 (0.8) |
| Torres Strait Islander | 1 (0.8) |
| Non-Indigenous | 127 (98.4) |
| **Education** | |
| Did not complete high school | 7 (5.4) |
| Completed high school | 7 (5.4) |
| Certificate | 22 (17.1) |
| Diploma | 23 (17.8) |
| Bachelor | 38 (29.5) |
| Masters | 22 (17.1) |
| Postgraduate Diploma | 10 (7.8) |
| **State or Territory** | |
| Australian Capital Territory | 12 (1.6) |
| New South Wales | 30 (23.3) |
| Queensland | 18 (14.0) |
| South Australia | 10 (7.8) |
| Tasmania | 12 (2.3) |
| Victoria | 54 (41.9) |
| Western Australia | 12 (9.3) |

| Demographics of respondents (n = 129) | N (%) |
|---|---|
| **Locality** | |
| Metropolitan or urban | 81 (62.8) |
| Regional | 41 (31.8) |
| Rural or remote | 7 (5.4) |
| **Language** | |
| English | 80 (62.0) |
| Non-English | 8 (6.2) |
| Missing | 41 (31.8) |
| **Disabilities, genetic and mental health conditions** | |
| People with a disability, genetic and/or mental health condition | 21 (16.3) |
| People without a disability, genetic and/or mental health condition | 108 (83.7) |
| **Relationship to child** | |
| Mother | 121(93.8) |
| Father | 6 (4.7) |
| Guardian | 2 (1.6) |
| **Number of children per household** | |
| 1 | 107 (82.9) |
| 2 | 16 (12.4) |
| 3 | 5 (3.9) |
| 4 | 1 (0.8) |

**Table 3: Demographics of survey respondents' children**

| Demographics of children (n = 158) | N (%) |
|---|---|
| **Child's age** | |
| Under 12 months | 12 (1.9) |
| 1 – 5 years | 40 (25.3) |
| 6 – 12 years | 74 (46.8) |
| 13 – 17 years | 28 (17.7) |
| 18+ years | 13 (8.2) |
| **Diagnostic status** | |
| Diagnosis confirmed | 102 (64.6) |
| No diagnosis | 56 (35.4) |

### 4.1.2. Interview participants

Eight interviews were conducted with parents of children with undiagnosed or rare genetic conditions, two of whom had two children with undiagnosed or rare genetic conditions. The demographics of interview participants and their children are shown in Table 4.

**Table 4: Demographics of interview participants**

| Demographics of interview participants (n=8) | N |
|---|---|
| **Gender** | |
| Female | 7 |
| Male | 1 |
| **State or Territory** | |
| Queensland | 1 |
| South Australia | 1 |
| Victoria | 5 |
| Western Australia | 1 |
| **Locality** | |
| Metropolitan or urban | 4 |
| Regional | 3 |
| Rural or remote | 1 |
| **Number of children with SWAN condition per household** | |
| 1 | 6 |
| 2 | 2 |
| **Child's age** | |
| 1 – 5 years | 4 |
| 6 – 12 years | 5 |
| 18+ years | 1 |
| **Child's diagnostic status** | |
| Diagnosis confirmed | 6 |
| No diagnosis | 4 |

Out of the 10 children of participants, 6 had received a diagnosis and 4 had not, one of whom was waiting on test results at the time of interview. Of those who had received a diagnosis some reported this occurred shortly after birth while for others it took several years.

Participants reported that seeking a diagnosis involved seeing numerous specialists and their child undergoing many tests and assessments, often resulting in hitting 'dead ends and roadblocks' with tests coming back negative and therefore having no explanation as to their child's condition and therefore their prognosis or likely support needs as they grow.

> *"We've done pretty much most things in New Zealand, Australia, England and in America that we can sort of do at this stage…. we might never know"*
>
> *(Parent of a child aged 6-12 years without a diagnosis, Queensland)*

> *"I've lost count of how many different tests we've done"*
>
> *(Parent of children 6-12 years and 1-5 years, with and without a diagnosis, Victoria)*

While some genetic testing is now covered by Medicare, participants reported they had experienced significant out of pocket costs on testing for their child.

### Description of Support Needs

The children of interview participants had varying impairments, from mild to severe, across all domains of function - physical, communication, vision, hearing, social-emotional, cognitive, self-care. Several also had specific health or medical needs. The level of support needs described ranged from one child needing specific nutrition-related support only, through to another who required 24/7 care and support for all aspects of daily life.

When asked about the types of supports needed for their child or family because of their child's condition, participants described the following groups of supports –

- Support workers: to assist in the day-to-day care of their child, for example bathing, toileting, feeding, and supporting access to recreational and extra-curricular activities such as swimming lessons or going to the park, to allow parents to manage competing demands of home, work, and education.
- Therapeutic supports: including occupational therapy, physiotherapy, psychology, speech pathology, and exercise physiology to support and facilitate independence and development of movement, communication, self, care, cognitive, and social-emotional skills.
- Assistive Technology: including mobility aids, eyeglasses, hearing aids, communication devices, continence products, orthoses, adaptive play and recreational equipment, car seating, vehicle modifications, equipment for toileting and bathing; some reported needing to hire equipment while waiting on long term provision or when on holidays.
- Transport: to travel to and from appointments, especially for those in rural and remote areas; additional transport costs to transport equipment when on holidays or other occasions when not at home.
- Home modifications:  to modify bathrooms, entrances, doorways to allow their child to access the home and safely manage self-care tasks.
- Support to access education: including modifying curriculum and teaching practices, flexible enrolment (e.g. dual enrolment), physical assistance, learning support, and support for social interaction with peers.
- Health/medical needs: these included attending specialist appointments, nutrition support and supplementation, respiratory support and medications.

## 4.2. Access to government funding schemes

Of the government funding schemes of interest in this study, nine (5.7%) of the children represented had not applied for any. Most participants had applied for the NDIS (n=139, 88%) and the Carer Allowance (n=113, 71.5%). The Pharmaceutical Allowance (n = 10, 6.3%) and Taxi Subsidy Scheme (n = 6, 3.80%) were the least applied for funding options (Table 5).

**Table 5: Funding schemes applied for**

| Funding Scheme | Applied for N (%) |
|---|---|
| **Carer Payment** | 57 (36.1) |
| **Carer Allowance** | 113 (71.5) |
| **Carer Adjustment payment** | 15 (9.5) |
| **NDIS** | 139 (88.0) |
| **Disability Support Pension*** | 17 (41.5) |
| **Education Support Supplement*** | 6 (14.6) |
| **Pharmaceutical Allowance** | 10 (6.3) |
| **Companion Card** | 56 (35.4) |
| **Taxi Subsidy Scheme** | 6 (3.8) |
| **WeCare Card**** | 19 (28.3) |
| **Essential Medical Equipment Payment** | 17 (11) |
| **Have not applied for any** | 9 (5.7) |

*These payments are only eligible for individuals greater than 16 years (n=41)

**This payment is only eligible for Victorians (n=67)

Almost all children (144, 91%) were receiving funding from at least one funding scheme. In line with application rates, the NDIS (136, 86%) and the Carer Allowance (101, 64%) were the schemes with the highest rates of respondents receiving them at the time of the survey (Table 6).

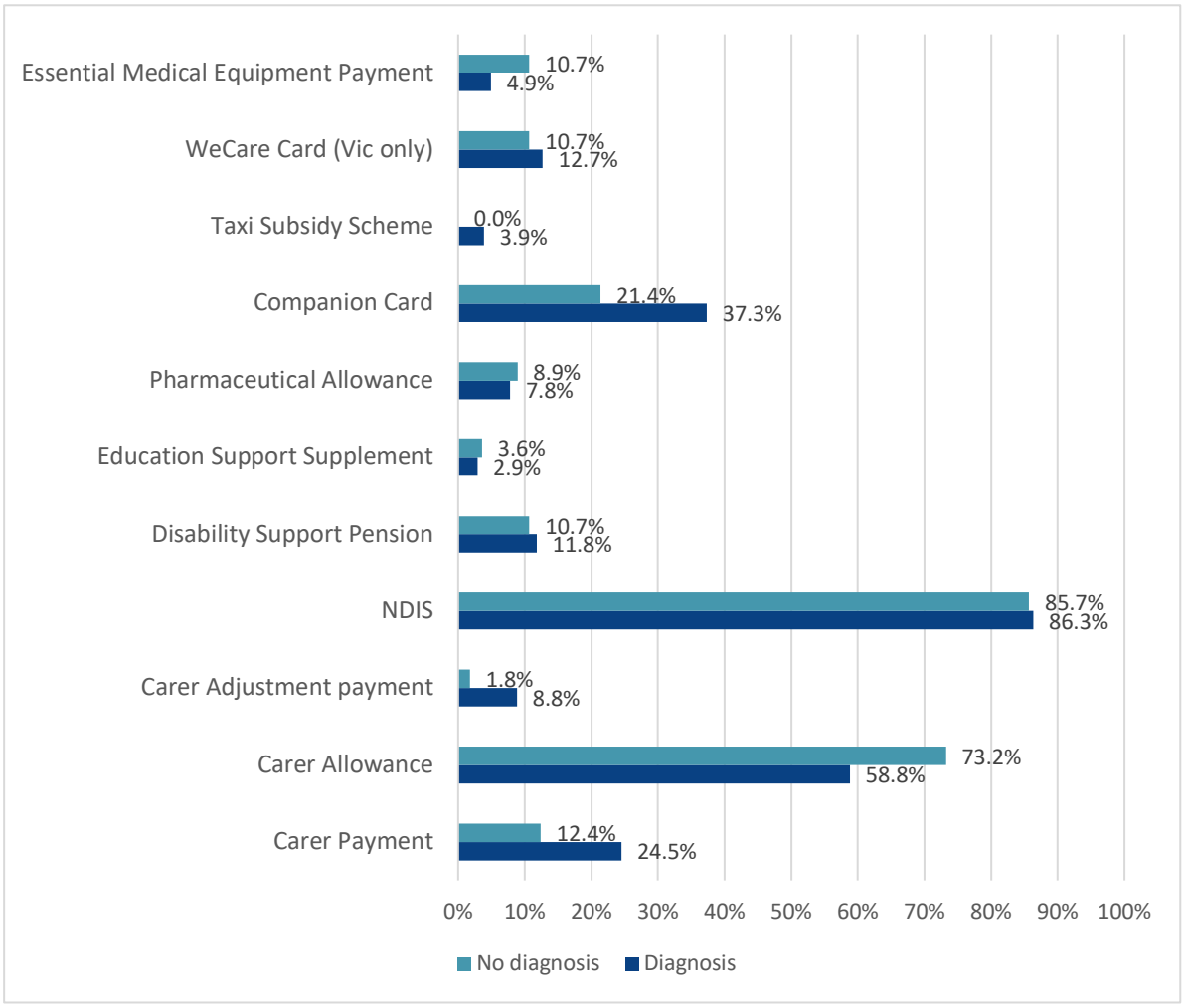
The main reasons respondents reported for never having received funding were 1) not meeting eligibility criteria (e.g. Carer Payment, Carer Allowance, NDIS and DSP), 2) because they did not know about the funding scheme (e.g. Carer Adjustment payment, ESS, Pharmaceutical Allowance, Companion Card, Taxi Subsidy Scheme, WeCare Card, Essential Medical Equipment Payment), or 3) the application process was too complicated (NDIS).

**Table 6: Funding received**

| Funding Scheme | Currently receiving N (%) | Never received N (%) |
|---|---|---|
| **Carer Payment** | 37 (23.4) | 90 (57.0) |
| **Carer Allowance** | 101 (64.0) | 30 (19.0) |
| **Carer Adjustment payment** | 10 (6.3) | 94 (59.5) |
| **NDIS** | 136 (86.1) | 11 (7.0) |
| **Disability Support Pension*** | 18 (43.9) | 19 (46.3) |
| **Education Support Supplement*** | 5 (12.2) | 19 (46.3) |
| **Pharmaceutical Allowance** | 13 (8.2) | 100 (63.3) |
| **Companion Card** | 50 (31.6) | 72 (45.6) |
| **Taxi Subsidy Scheme** | 4 (2.5) | 108 (68.4) |
| **WeCare Card**** | 19 (28.3) | 31 (46.3) |
| **Essential Medical Equipment Payment** | 11 (7) | 99 (62.7) |

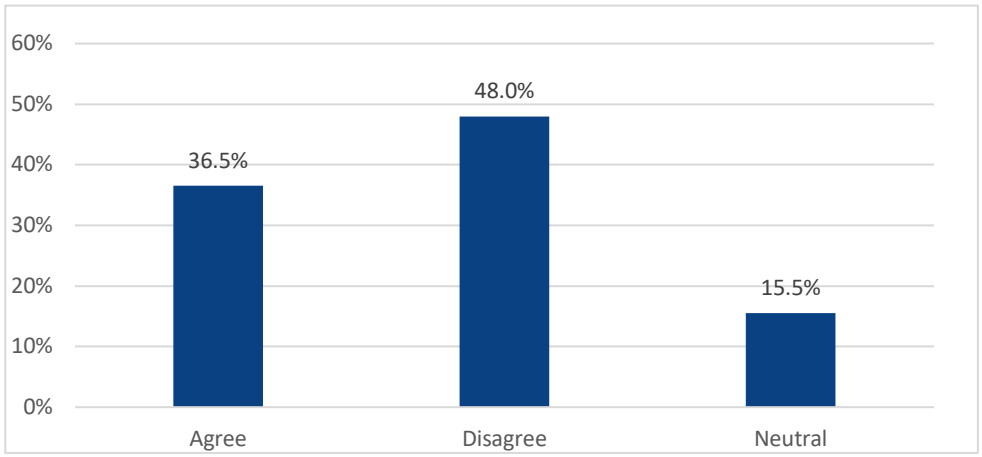
When comparing access to funding schemes for children with and without a confirmed genetic diagnosis (Figure 1) the percentage of children currently receiving NDIS funding was approximately the same for both groups (diagnosis 86.3%, no diagnosis 85.7%), as was access to the WeCare Card, Pharmaceutical Allowance, Education Support Supplement, and Disability Support Pension. For the Carer Payment, Carer Adjustment Payment, and Companion Card the percentage of children with a diagnosis currently accessing those schemes was higher than those without a diagnosis. Whereas access to the Carer Allowance and Essential Medical Equipment Payment was higher among those without a diagnosis compared to those with a diagnosis.

**Figure 1: Children without a diagnosis currently receiving funding compared to children with a diagnosis**




Of those who applied for funding (n= 148), almost half of respondents (n=71, 48%) reported their child's overall funding amount was insufficient to support their needs, while 36.5% (n=54) of respondents believed they received sufficient funding (Figure 2).

**Figure 2: Overall funding currently received is sufficient to meet child's support needs**



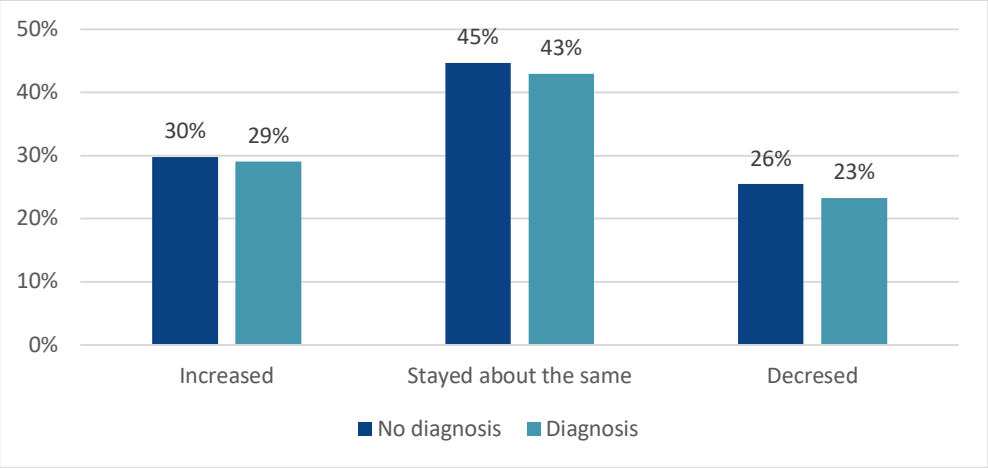

Almost half of respondents whose children were receiving funding from the NDIS reported that their child's current NDIS plan was insufficient to meet their support needs (n=61, 47%). This was slightly higher for

individuals with a confirmed genetic diagnosis (n=41, 47% ) compared to those without a diagnosis (n=20, 42%).

Some participants reported experiencing reductions in the funding they received. These included NDIS (n=47), Carer Payment (n=10), Carer Allowance (n=7), and Disability Support Pension (n=3). The most cited reason for reductions in the Carer Payment or Carer Allowance was increased household income.

Among those with NDIS funding, approximately one third (n=39, 29.3%) reported that their most recent plan had increased compared to the previous plan, while a quarter (n=32, 24.1%) reported their plans had decreased, and a little under a half (n=58, 43.5%) had plans that remained about the same. There was little difference between those with and without a diagnosis (Figure 3).

**Figure 3: NDIS plan amount in current plan compared to previous plan**




Of those who had NDIS funding reduced, the key perceived reasons for this were the NDIA's lack of knowledge and understanding of undiagnosed and rare conditions; their child had turned 7 and was no longer eligible for early childhood early intervention (ECEI) supports; and unclear application and administrative processes.

## 4.3. Barriers and enablers of access to government funding schemes

### 4.3.1. Barriers of access to funding

#### Quantitative findings

Survey respondents were asked about the main challenges they faced when applying for and accessing government funding schemes. Complex application processes were cited as a key barrier for many respondents (n=66, 41.8%). Respondents indicated that Carer Payment, Carer Allowance, Carer Adjustment Payment, NDIS, and DSP were difficult to apply for. The reasons given for these application difficulties included lengthy paperwork, requirements for extensive supporting documents, long wait times for funding outcomes, and multiple attempts required before they were approved for funding. One respondent said that *"18 pages with a medically complex newborn is hard work not to mention having to get multiple sign offs from medical practitioners".* Another parent reported that her child passed away while waiting for the outcome of their funding application.

WeCare Card and Essential Medical Equipment payment were the only government funds that respondents reported as being easy to apply for.

Aside from the application process, several barriers to accessing funding were reported (Table 7), including a perceived lack of understanding from staff assessing funding applications (n=82, 51.9%), lack of knowledge by parents on available funding schemes (n=79, 50%), a lack of recognition of undiagnosed or

rare conditions under various government funding schemes (n=68, 43%), and costs of obtaining necessary documentation (n=36, 22%).

**Table 7: Key challenges with accessing government funding**

| Challenges | N (%) |
|---|---|
| **Lack of understanding by staff assessing funding e.g. Centrelink, NDIS Planners, Local Area Coordinator (LAC), Early Childhood Approach Partners** | 82 (51.9) |
| **Lack of knowledge of available funding support schemes** | 79 (50.0) |
| **Child's condition not recognised under the scheme/s** | 68 (43.0) |
| **Complex application processes** | 66 (41.8) |
| **Lack of diagnosis** | 66 (41.8) |
| **Cost of obtaining supporting documents** | 36 (22.8) |

Age related factors were also reported as barriers, with several participants reporting reduced NDIS funding once their child turned 7 years of age, and again when children turn 16 years with one respondent stating - *"there are challenges once the child turns 16. There is no smooth transition and often payments are cancelled".*

Respondents of children approaching the age of 7 were worried that funding was more likely to be reduced at that point if their child did not have a diagnosis.

Lack of diagnosis or having a rare diagnosis were considered to be key barriers. Over two-thirds of respondents (69.0%, n=89) reported believing that accessing government funding for children with undiagnosed and rare genetic conditions was not the same as for children with more well-known conditions such as autism and Down Syndrome.

More than half of respondents (52.7%, n=68) believed NDIS plans for children with undiagnosed and rare genetic conditions were not equivalent to those of children with more well-known conditions (e.g. Autism, Down Syndrome) with similar support needs. Respondents perceived Local Area Coordination (LAC) staff's lack of awareness and understanding of the impacts of undiagnosed or rare conditions, along with inexperience in assessing for their needs, may have contributed to different NDIS funding plans for those with more well-known conditions. A survey respondent stated

> *"I have a child with a well-known condition and two with rare conditions and the plans are worlds apart. I am also friends with parents of those with well-known conditions and it seems to be easier to put them [applications] through, less paperwork and fighting".*

### Qualitative findings

Interview participants described similar barriers to those raised by survey respondents. These included complex or unclear processes, lack of awareness of funding schemes, or being deemed ineligible.

Almost all interview participants discussed challenges when applying for funding support, in particular when applying for the NDIS or the Disability Support Pension.

Both the initial application and planning review processes for the NDIS were described by participants as being confusing, complex and arduous. Several reported that they found the application process complicated and difficult to understand and wondered how people with lower levels of education, literacy or support would be able to navigate it.

*"I feel for those people that don't have the support networks, that don't have the organizational skills, and don't have the intellect to be able to deal with these systems because it's absolute rubbish. And I feel for the poor girl that has been through a bad separation, whose mum has passed away, and whose child is kicking the crap out of the walls, and she doesn't know where to go, what services are available to her. Because she can't read properly, she can't understand what someone's asking her to do, and the 85 pages of documentation that they ask you to submit."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

*"I'm relatively educated. I work for the government. You know, I've got a good job. I've done business analysis and like stuff like that. I have very good comprehension of the English language and I still can't figure half this stuff out and I actually feel for the people that don't or can't speak English well or that just have, literacy problems or a disability themselves that they're trying to navigate the system."*
*(Parent of a child aged 1-5 years without a diagnosis, Victoria)*

Participants reported that obtaining the necessary reports and documentation for applications and plan reviews required significant time, effort, and, often, cost. They discussed long waiting lists to obtain reports through the public system but significant, often unaffordable, costs of seeking these privately.

Some participants reported once their applications had been submitted that they were plagued by systemic issues with applications going missing and needing to re-submit or having to wait a long time before receiving an outcome. They also reported that due to high turnover of NDIS staff they rarely had the same person twice for plan reviews, and believed the process and outcome was dependent on the skill and knowledge of the individual planner.

*"We've been submitting the same stuff for the last two years; this is so dependent on who sees it and who reads it and what channel it happens to go through."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

One parent reported that after going through the NDIS application process for one of her children she could not face going through it again for her other child even though they have significant support needs and is was confident they would be eligible -

*"The navigation of the [NDIS] system is too hard. I now understand it having worked through six years of it, but it's been a hard road and anybody that isn't educated, it's almost impossible to navigate."*
*(Parent of children aged 6-12 years and 1-5 years with a diagnosis, Western Australia)*

The one participant whose child had applied for the DSP described the process as 'awful', and that similar to the NDIS it required lots of assessments and reports, and they experienced difficulties with applications going missing and long waiting times when attempting to apply in person.

Many of the participants reported that they had not heard of some of the available funding schemes or only found out after they were told about funding for another of their children with a more well-known diagnosis. They felt that without a diagnosis they were less likely to receive information, and without prior experience of the disability support system, they, in a sense, didn't know what they didn't know, so it didn't occur to them to seek it out.

*"So it's all quite new. And my ex-wife is the same, we haven't had the need for [funding support] before. So, you're not aware of what's available and the process involved in getting it."*
*(Parent of children 6-12 years and 1-5 years, with and without a diagnosis, Victoria)*

One participant who worked as a disability advocate said that even she was still learning of funding that her child was eligible for after many years —

> *"I'm an advocate and I didn't even know"*
> *(Parent of a young person over 18 years with a diagnosis, Victoria)*

Being deemed ineligible was the other main barrier to accessing funding raised in interviews. The two reasons cited by participants for being ineligible were due to household income for schemes that are means tested, or on the basis of their child's rare diagnosis or lack of diagnosis.

Of those who were not eligible for the carer payment due to income, some reported that this impacted their decisions as to whether to work or not. Working part time could be enough to lose the payment but working part time was also not enough to meet household expenses, so they had to choose between full-time working or full-time caring.

One participant reported being rejected for the carer allowance on the basis that caring for a 3-year-old was deemed 'parental responsibility', despite their child's high support needs. The participant reapplied twice and was successful on their third attempt with a 'strongly worded letter' from their paediatrician.

A lack of diagnosis or having a rare genetic diagnosis were reported as a key barrier to funding as participants felt eligibility or level of funding was more easily determined with a diagnosis, and more so with a diagnosis of a relatively well-known condition.

Many parents described that their child's needs are difficult to categorize or prove during application processes without a diagnosis. As one parent said -

> *"It's hard to write down what she has because effectively she doesn't have anything, she actually has no diagnosis, but she has a lot of things on the other hand. All of her negative tests that we've done are a real negative now when it comes to filling out paperwork because you answer all those questions as no, she doesn't have any of those things [diagnoses]."*
> *(Parent of a child aged 6-12 years without a diagnosis, Queensland)*

Comments of needing to "jump through hoops" to explain their child's needs or that their child "doesn't fit in any of the boxes" were common. Parents reported that they understood the NDIS had pre-determined categories and when their child's needs didn't neatly fit these they were deemed in-eligible or given funding that did not appropriately meet their needs.

> *"So whenever you're trying to access something, the first thing you get handed is a form. And we love to fit things into a box or tick a box, and when you don't have a diagnosis and a prognosis, it's really hard to fit into that, and then once that form goes in and you enter into that programme or that system they're not quite sure how to handle him because he doesn't have a diagnosis in there. He sort of doesn't fit into a box."*
> *(Parent of children 6-12 years and 1-5 years, with and without a diagnosis, Victoria)*

> *"The NDIA doesn't really seem to have a kind of a complex, rare needs type box or take into consideration when there's so many different factors."*
> *(Parent of a child aged 1-5 years without a diagnosis, South Australia)*

Even for those with a diagnosis, there are still challenges in demonstrating their child's support needs and therefore eligibility, due to the lack of information available about the condition and often unknown prognosis. One parent reported that their child was only the 8$^{th}$ person in the world to have been diagnosed with their condition, a condition which had only been documented in medical literature for the first time 12 months before her child's diagnosis. Consequently, there is a lack of knowledge and evidence as yet about the condition to support eligibility for funding.

*"We don't have evidence for why he needs these things because he's rare, because there are not research papers, there's not treatment protocols and things around it."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

*"I don't think they could get their head around the complex rare needs that he has because it is so unusual, his unique combination of needs is not comparable to anything."*
*(Parent of a child aged 1-5 years without a diagnosis, South Australia)*

Most participants reported that their child was not present at NDIS plan reviews and that Planners had never met their child and relied on reports and information from parents to understand their functioning and support needs.

Other barriers related to having difficulty demonstrating support needs for conditions that fluctuate or have periodic 'flare ups' and genetic conditions being viewed as a health condition and therefore not eligible for disability-related funding.

*"But the problem is genetic conditions they [NDIS] see fits in health. And that's where they keep handballing it. So we've got all these families whose kids need support missing out. Even though you might have all these reports to say that this child is quite complex and the impact on the carer you know, is very complex as well. The NDIS is just it's just all too hard. Families are just getting told no, no, no."*
*(Parent of a young person over 18 years with a diagnosis, Victoria)*

Several participants with children under 7 described that without a diagnosis they feared their child would no longer be eligible for NDIS funding once they turned 7, with some reporting having been advised that they were very likely to have their funding cut.

### 4.3.2. Enablers of access to funding

#### Quantitative findings

Survey participants were asked about key sources of information or support that assisted them when applying for and accessing government funded supports. These were reported as allied health professionals, their child's paediatrician and/or other medical specialist, other parents, social media, their NDIS support co-ordinator and advocacy organisations. Multiple respondents in the study also found self-research assisted with accessing government funding.

Survey participants were also asked to identify key strategies or actions that they believed enabled success in accessing funding schemes. These included having reports of their child's functioning and support needs (n=115, 89%), having diagnostic assessment reports (n=90, 70%), contacting their local member of Parliament or other government representative (n=24, 19%), or having a professional advocate (n=16, 12%).

Appealing decisions or resubmitting applications with additional information were also reported as enablers for NDIS funding access. Those whose child's most recent NDIS plan had increased compared to previous plans reported they believed this was due to submission of additional evidence and escalation of their plan to a higher delegate for review.

#### Qualitative findings

Interview participants discussed many enabling factors that assisted them to access government funding support for their child. These were mostly discussed in relation to accessing and maintaining NDIS plans, and could be broadly grouped into 1) support from health professionals; 2) strategies implemented by the family; 3) funding scheme factors; and 4) seeking additional support.

Almost all participants reported receiving information about funding schemes, encouragement to apply, and supporting reports and other documentation from their child's treating health professionals, and that this was key to accessing funding.

*"When he was in hospital, we were really lucky that a social worker told me about the carer adjustment payment."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Having health professionals who had a good knowledge of the NDIS and what documentation was required was reported as particularly helpful. In some instances where applications were initially unsuccessful, participants found that obtaining further specialist assessments and reports as evidence facilitated access.

*"So basically we had to go to the next level of evidence basically to get us over the mark. Letters from the Children's hospital, letters from professors at the Children's, letters just basically providing more evidence to say yes the child is not 'normal'."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Factors relating to family resources that participants believed enabled access included having the financial capacity to pay for additional assessments and testing privately, and having a family member with a health or disability background that could assist with navigating the system, such as for one family where the child's mother is a social worker -

*"I think a lot of that just comes down to having someone in our family [social worker] that knows how to write a plan, and knows the lingo or terminology."*
*(Parent of children 6-12 years and 1-5 years, with and without a diagnosis, Victoria)*

However, even those with relevant knowledge found the application processes challenging and time-consuming.

Participants also reported they used their own strategies such as providing detailed carer statements, comparing their child's need to those of a typically developing child, and choosing not to ask for certain supports for fear of "rocking the boat" and losing the funding they already had.

*"We just haven't rocked the boat. I definitely wouldn't go out of my way to ask for more in the sense that we might lose funding if I ask for more. So I'd just be happy to keep what we've got."*
*(Parent of a child aged 6-12 years without a diagnosis, Queensland)*

Seeking support and information from other families of children with rare and undiagnosed genetic conditions about the strategies they used to access funding was a further strategy used by several participants.

*"I have spoken to other mums on Facebook and all the socials to try and find out how they got their packages. I've reached out to people that I don't even know."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

For NDIS funding, enabling factors included having access to a Support Co-ordinator to assist with compiling evidence for plan reviews and emotional support, and having a knowledgeable and experienced LAC/ECEI Partner or NDIS Planner.

*"I picked up a really good planner, and I went into that meeting with, like the same evidence that I had always had, but with gung ho. And she was fantastic. We had her again for our next plan review. It was the first time I've probably gone in with relief like, not stressed to the max in a meeting because we still had her again."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Appealing funding decisions or requesting reviews of funding decisions was a further strategy some participants employed to access funding. Using an advocate to support appeals processes, and writing to Members of Parliament were also reported as helpful.

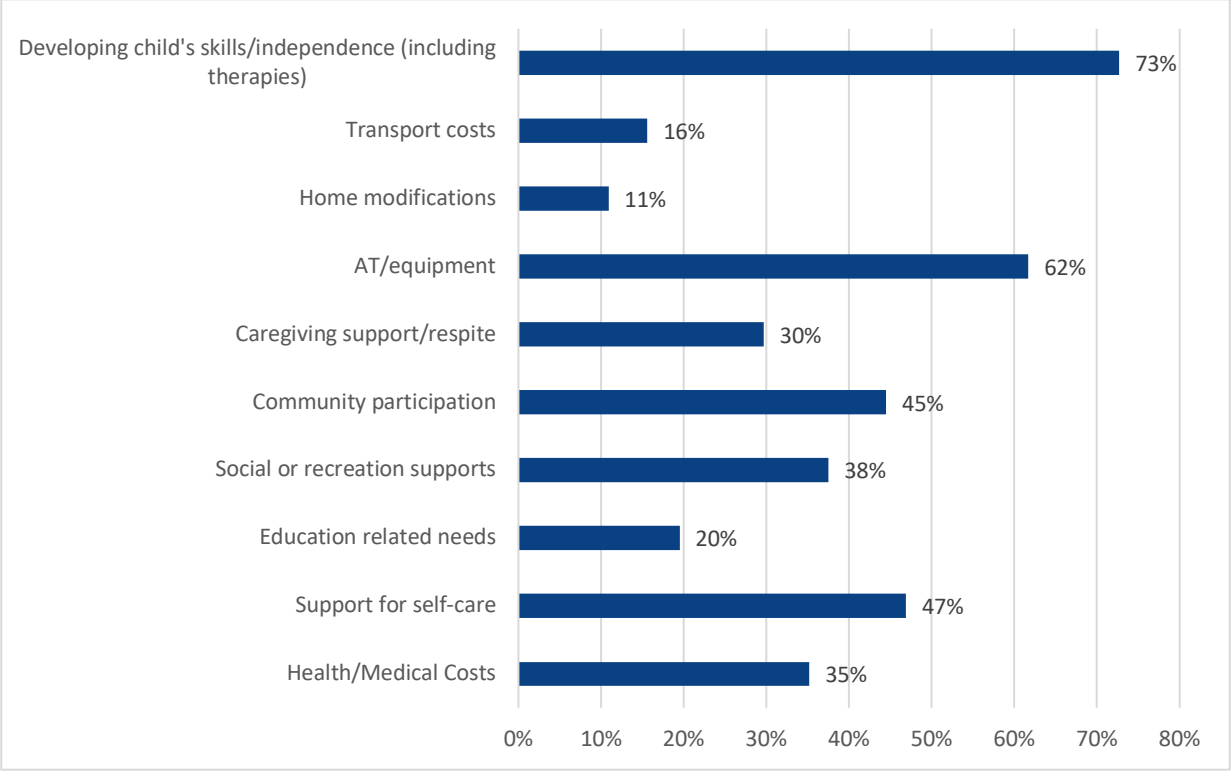
*"I ended up calling the local member of parliament and going down that avenue for them to chase it to say, Hey, we haven't got anything what's going on? That kicked it on and we got a plan that was lots better than the one before."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

## 4.4. Use of funding and positive impacts

### 4.4.1. How participants use funding they receive

Survey respondents were asked about how they use the funding they receive from government funding schemes (Figure 4). Nearly three quarters of respondents reported using funding to assist in developing their child's skills and independence (n=93, 73%), and more than half used funding to access assistive technology (AT) or other equipment for their child (n=79, 62%). Almost half use funding for supporting their child's self-care needs (e.g. bathing, toileting) (n=60, 47%) and community participation (n=57, 45%), while about a third use funding for caregiving support/respite (n=38, 30%), social or recreational support (n=48, 38%), and to cover health care or medical costs (n=45, 35%).

**Figure 4: How funding is used**




Interview participants similarly described that they used funding received from government funding schemes to access therapy services (e.g. occupational therapy, speech pathology, physiotherapy, play therapy, dieticians), purchase assistive technology (e.g. mobility and communication devices, specialist feeding equipment, vehicle modifications) and fund home modifications. They also discussed using funding to access support workers to assist with their child's personal care and participating in recreational activities such as Auskick, swimming lessons and outings to the zoo or park, enabling parents time to undertake other household or work activities.

*"We get a support worker to take [child] out to do stuff. So taking her to like the zoo or the park. And I actually use that time when she's gone to clean, mow the lawn, or you know I go to the supermarket with my massive list of shopping list because getting her in and out of the car and the walker in and out of the car and finding a spot that works is just so time consuming. It's hard to manage a walker and a trolley at the same time because I still*

*have to help her move in the right direction. So we kind of either now have to do click and collect or do it when she's not around, which is hard because she's always around."*
*(Parent of a child aged 1-5 years without a diagnosis, Victoria)*

Some also reported using funding, from the Carer Allowance for example, to help pay for medications or fuel to attend regular appointments for their child.

#### 4.4.2. Positive impacts of access to funding

Interview participants discussed some of the positive outcomes or impacts for their child and family as a result of having access to government funding.

Several participants reported that NDIS funding enabled access to therapies and assistive technology that was 'life changing', seeing significant improvements in their child's functioning and wellbeing as a result. They described progress in their child's communication and motor skills, in particular.

*"It [communication device] was the best thing for her at the time, but now she doesn't even use a device because she has enough words. She can communicate effectively enough now. It's wonderful seeing her have a conversation with the kids at school and the teachers, where she didn't have that previously."*
*(Parent of a child aged 6-12 years without a diagnosis, Queensland)*

*"His first signs, his first ability to crawl and step, these things have all happened in those [NDIS funded] therapy blocks."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Some discussed that by having access to funding, they were able to prioritise their own money for other things that can't be funded through government funding but were also important for their child's or family's wellbeing, such as holidays or other recreational activities.

One participant stated that access to funding meant she no longer had to undertake fundraising activities to fund her child's therapies and equipment, while another commented that access to the NDIS had *"changed our life, financially, enormously."*

Access to funding for support workers was a key benefit discussed by several participants. They described that being able to have support workers enabled parents to work that could not previously, and helped families to manage the varying household needs and activities of their children with disabilities and their siblings.

*"Support workers are paramount to keeping us floating above water, to keeping our marriages stable, to keeping siblings from being left out. If [child] is having a bad day with seizures, we can use support workers to help us manage so that [sibling] does not have to miss out on going swimming or to a birthday party, or so that we can work. That has made our world liveable, bearable and we're functioning, and we're still valuable members of the community. We're still contributing in terms of the business sense and everything else."*
(Parent of children aged 6-12 years and 1-5 years with a diagnosis, Western Australia)

Some participants also reported the flexibility of NDIS funding compared to previous schemes, enabled them to choose their own service providers, and this allowed them to find someone that could work around their work and school schedules and that this was extremely beneficial.

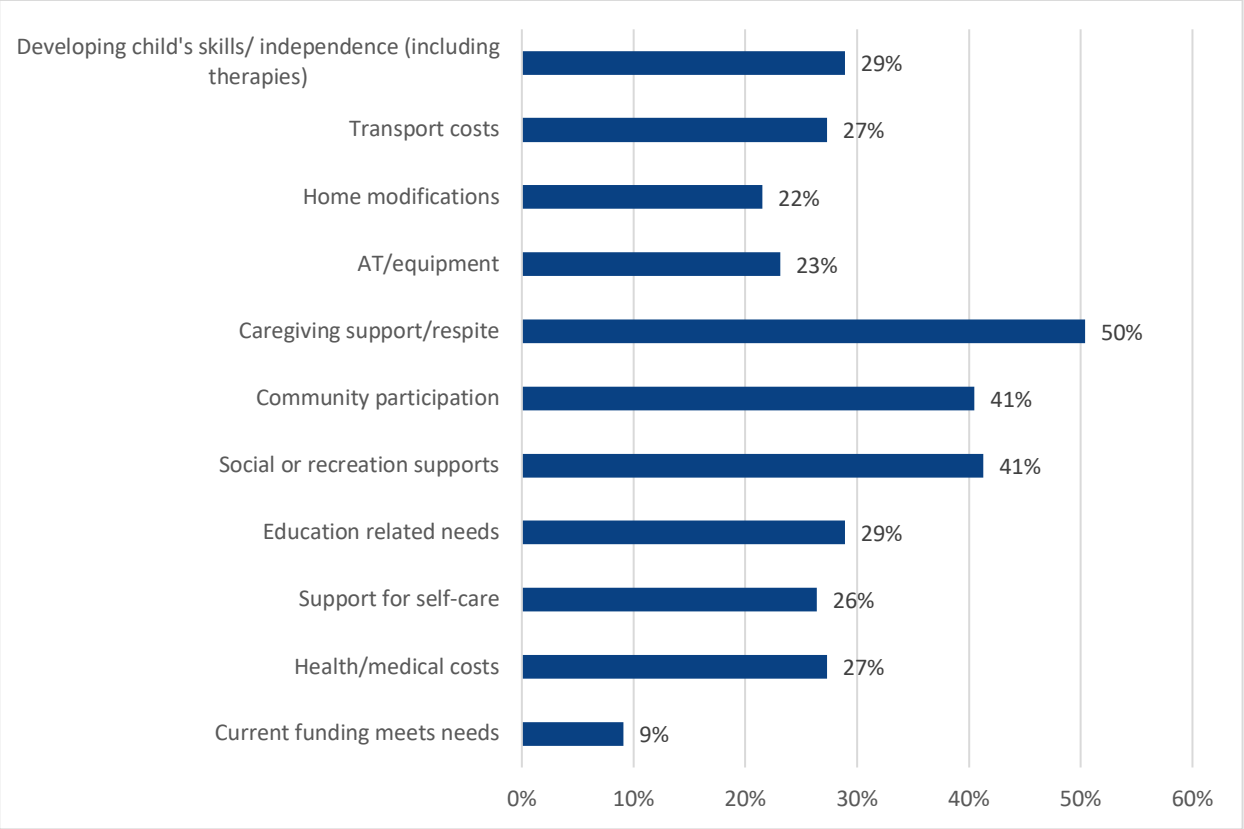
### 4.5. Funding gaps and impact of unmet support needs

#### 4.5.1. Unmet support needs

Of those survey respondents who reported that current funding was insufficient to meet their child's support needs, more than half (n=40, 56%) stated that as a result their child goes without needed supports.

The highest unmet need was for caregiving support, or respite, with half (n=61, 50%) of all respondents report being unable to meet this need. Community participation (n=49, 41%) and social or recreational support (n=50, 41%) were the next highest unmet need. Less than ten percent (n=11) of participants reported that their current funding level meets their child's support needs.

**Figure 5: Unmet needs with current funding**




Interview participants also reported similar unmet needs, with insufficient funding for support workers to assist in providing care and support for their child being their greatest unmet need. This included support for assisting their child to complete self-care tasks, to accompany their child on social and recreational activities, or to accompany the family on outings so that there was adequate supervision and support for their child's support needs and that of their siblings –

> *"Even if we had three hours of support work so I could take them [children] somewhere, and that carer would help me do what [child with disability] wanted to do, because otherwise you have to follow him around, I don't get any time with my other child."*
> *(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Participants also discussed a short-fall in funding for continence related products for their child and difficulty accessing funding to upgrade AT to accommodate their child's growth or changing needs and having to seek funding through other means.

> *"We wouldn't have had anything to transport him in unless we approached a charity. He's too big for his wheelchair, he's had the chair since he was two, it's literally falling apart."*
> *(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Insufficient funding for behaviour supports, communication devices, and to support access to recreation (e.g. private swimming lessons) were also reported gaps.

### 4.5.2. Strategies used by families to manage funding gaps

Most survey respondents with insufficient access to funding reported attempting to manage this shortfall through paying for supports out of their own pocket (n=56, 78%). This included utilising their personal long-

term savings (n=43, 60%) borrowing money from family or friends (n=31, 44%), taking out personal loans (n=4, 5%), or refinancing their mortgage (n=4, 5%). Some were able to use private health insurance to help meet funding gaps (n=26, 37%), others reported approaching charities (n= 17, 24%), or reducing work hours so they could provide the support themselves (n=3, 4%).

Interview participants similarly discussed using their own money to meet funding gaps, with some reporting taking on additional work hours to do so.

> *"You have to find the extra $1000 to try and get nappies and to get the extra pads and to get the all the bits that we need."*
> *(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

> *"My husband and I worked our guts out just to pay for [child's needs]."*
> *(Parent of a young person over 18 years with a diagnosis, Victoria)*

One parent reported they had to cover approximately a quarter of support costs themselves, while acknowledging that this is not an option available to everyone.

> *"I would say we're probably about 75% funded, so we might be like 20 or 25% out of pocket. So yeah, that's probably where we sit at the moment with the NDIS. We've just paid for that [gap] ourselves to keep her funding for her therapies. But a lot of people would have missed out on being able to have those services because they wouldn't be able to afford them."*
> *(Parent of a child aged 6-12 years without a diagnosis, Queensland)*

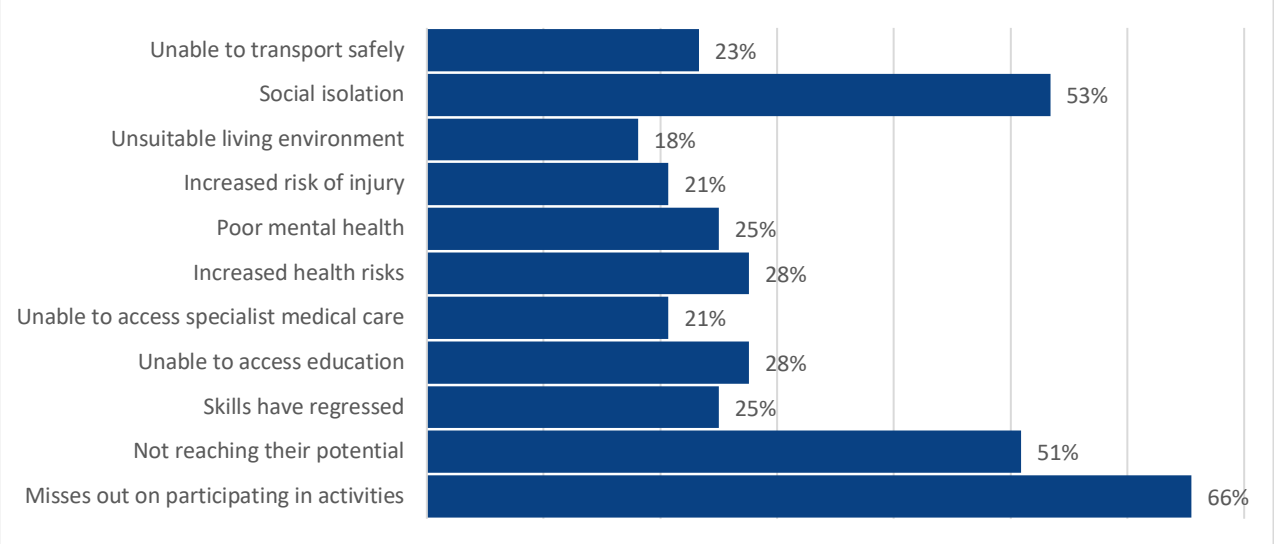
Participants knew of families contemplating selling their homes to fund the cost of their child's support needs, and as for survey respondents, some interview participants approached charitable organisations for funding, including the Steve Waugh Foundation, Rotary, and TLC for Kids. One parent reported conducting their own fundraising activities to pay for AT costs.

### 4.5.3. Impact of lack of access to government funding

#### Impacts for children

When asked about how insufficient access to funding impacts on their children, more than half of survey respondents identified that it means their child misses out on participating in activities (n=76, 66%), that they are not developing skills and reaching their potential (n=59, 51%) and that their child is socially isolated (n=62, 53%).

**Figure 6: Impact for child of lack of access to sufficient funding**

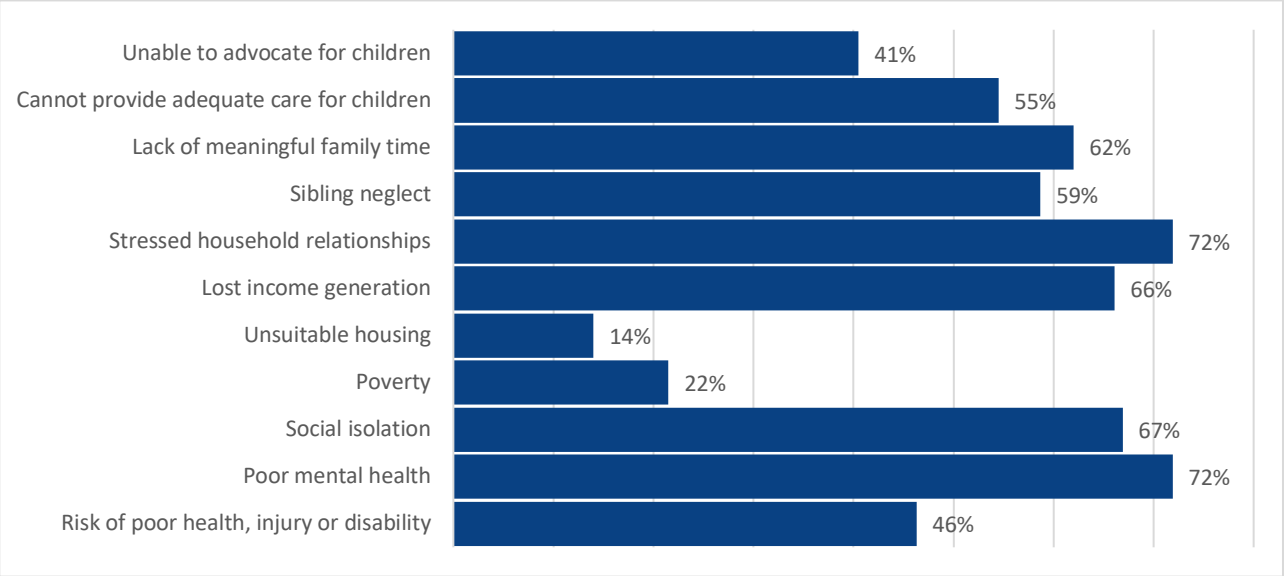


These findings were supported by the findings from the qualitative interviews in which participants also discussed the main impact on their children as being a lack of opportunities for social participation due to not having appropriate AT, lack of support worker time, or having not had the therapy inputs to support implementing strategies for social participation.

### Impacts for parents/caregivers and families

Survey respondents with insufficient access to government support for their child also reported many significant impacts on themselves and their family more broadly (Figure 7). Almost three quarters reported poor parent/caregiver mental health (n=87, 72%) and stressed household relationships (n=87, 72%). Approximately two thirds also reported experiencing social isolation (n=81, 67%), lost income generation opportunities (n=80, 66%), and lack of meaningful family time (n=75, 62%). Feeling unable to adequately care for their children, both those with disabilities and their siblings was also a concern of the majority of respondents. One respondent shared: *"I worry that our family will fall apart from the weight of the huge burden and stress. I also worry about the physical and mental health on both of us [parents] in our caring roles, the stress that comes from having to fight the system to get our daughter the supports she needs".*

**Figure 7: Impact on parents/guardians and families of lack of access to sufficient funding**




Interview participants particularly described impacts relating to financial stress, current and future caregiving pressures, and the impact on household relationships.

Participants reported reducing household spending to meet their child's disability-related support needs.

> *"We can't go on holidays because any extra money that we earn as a family or anything that we would put into holiday fund or savings or anything like that has to go towards wipes, extra clothes, bedding due to [child's] incontinence. We're at the moment trying to save up for a new mattress because the mattress is so just ruined from all of the urine. Mattresses get ruined, pillows get ruined. But no, they won't fund those, so we forego a lot of extra family type of things."*
> *(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

Without access to sufficient funding for support workers or respite, participants described the toll of providing all the care and support themselves, including reducing their employment (and therefore their superannuation) and using up leave entitlements such as long service leave.

> *"We are doing a lot of the informal support. But we're both in our sixties, and he [father] needs double knee replacement. We can't continue to do this alone anymore."*
> *(Parent of a young person over 18 years with a diagnosis, Victoria)*

*"We'd maxed out all our all our leave basically. My husband resigned from his role, which was a senior management role, basically because [child's] needs are complex and it means that sometimes we've got to drop what we're doing at very short notice. Because we've not got NDIS funding for support workers we didn't really know what else we were going to do."*
*(Parent of a child aged 1-5 years without a diagnosis, South Australia)*

They worried about the impact the lack of support has on their other children and their ability to do activities as a family.

*"So we can't go to the movies without help. We can't because [child] wants to walk off halfway through. We can't go to the zoo. We can't go because [child] wants to shoot off and look at something that's not of Sam's interest level."*
(Parent of a child aged 6-12 years with a diagnosis, Victoria)

And also reported the stress of the ongoing cycle of applying for funding and planning for their child's future support needs.

*"The repercussions of the stress that goes with the constant reapplication and budgeting and proving that your child's deficits are so great. You probably spend five months of the year stressed about your next NDIS plan….I just want to cry, on the way to work. I want to exercise for myself so that I am physically able to care for him and to be alive and work and be around him, and for me. But by the time I add that in that I can't have him ready for school. I'm having to carry out all the care and then I'm having to go to work, and I go to work because we need funds to pay for our housing. I need something for him to live in when he's 18. It's all the future implications of caring for him throughout his adulthood that these things need to happen."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

### 4.6. Suggestions for improvement

Interview participants were asked about what would improve the experience of accessing government funding for children with undiagnosed and rare conditions.

Many of the responses related to the NDIS but some were more general. These more general suggestions included –

- Raising awareness of the government funding available for children with undiagnosed and rare conditions so that families know what they can apply for.
- Developing peer support networks for navigating the processes when applying for and using funding.
- Training and support for families in managing funding to help them get the best value out of what is available.
- Making the eligibility for funding based on support needs rather than diagnosis.

*"I don't understand why her functional difficulties can't be taken into account and she can be boxed into another group just based on the disability rather than a diagnosis of a disability. I know, kids with cerebral palsy that are very, very, very mild that you probably wouldn't necessarily think they are disabled, but by default, because they've got a defined diagnosis get a lot different and a lot more funding."*
*(Parent of a child aged 1-5 years without a diagnosis, Victoria)*

Suggestions for improvement related to accessing NDIS funding included –

- NDIS Planners or delegates meeting the child and family face to face in their home environment to better understand their context and support needs.

*"Meet these families, go out and see the pressure these kids put on not only families but siblings and all of that sort of stuff. Every element of life, walk out and see them. Don't expect these parents to come to you because we can't prove everything. It's not possible because you can't put it on paper."*
*(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

- Support for navigating the application and planning processes for those who need or want it.
- Improve training of NDIS staff reading applications and plan review documents to better understand the information provided that demonstrates the needs of children with undiagnosed and rare conditions and improve consistency of funding decisions.
- For the NDIS to show greater trust in the evidence that is provided by families, including professional reports and their own description of their child's support needs, coupled with an understanding of the lack of available peer reviewed evidence on the prognosis and support needs for these children due to the rare nature of their conditions.

  *"There's not a large body of written evidence around these things. And there never will be. That parents, often with rare conditions, are being pretty honest about the levels of care. So ways to actually assess the care and not to just put it on to, like to have some comparison between what is typically expected of a child."*
  *(Parent of a child aged 6-12 years with a diagnosis, Victoria)*

- More consistency and less frequent changes to the requirements when applying for NDIS or undergoing plan reviews.
- To have a focal point within the NDIA for managing cases of children with rare or undiagnosed conditions who understands their unique circumstances especially around lack of extensive evidence on prognosis and outcomes, who can be a trusted point of contact and referral.

  *"I just think it needs to be a separate niche within NDIS. if we could refer someone to a really qualified quality delegate that really gets this and will look at it, you know, with open mind and eyes and see those reports of functional capacity."*
  *(Parent of a young person over 18 years with a diagnosis, Victoria)*

## 5. Discussion

Our study found that while most participants were receiving funding from at least one of the available government funding schemes, approximately half reported that it is insufficient to meet their child's support needs, resulting in significant out of pocket costs and unmet needs for children with undiagnosed and rare genetic conditions and their families. This is consistent with previous studies that also reported parents of children with rare diseases who had received insufficient funding to meet their needs [15, 17, 27, 28]. Further, there was a perception among participants in our study that funding received by those with undiagnosed and rare diseases is not equivalent to those with more common conditions.

### Barriers

Insufficient access to funding is due to a number of key barriers which include 1) lack of knowledge of available funding schemes; 2) difficulty demonstrating eligibility due to lack of diagnosis or having a rare diagnosis; and 3) systemic issues in the administration of funding schemes including complex application processes and lack of knowledge of staff of the needs of this cohort.

While awareness of some schemes, such as the NDIS and Carer Allowance, was high amongst participants, other schemes were less well known. Mechanisms for receiving information about the available funding schemes was variable but relied heavily on the individual knowledge of treating medical and allied health professionals to share this information with families or knowledge obtained through online support groups. Some reported that it was "luck" that led them to learn about funding schemes. It is possible that not having a diagnosis means individuals are less likely to be linked into support groups or networks where such information may be shared and treating health teams may be less likely to prompt applications for funding support.

Having a rare diagnosis, or not having a diagnosis, is a significant barrier to meeting eligibility for some funding support schemes and creates a high level of uncertainty and unmet support needs [8, 9, 25]. Families of children with undiagnosed and rare conditions face high levels of stress and uncertainty both with regards to obtaining a diagnosis to provide some understanding of their child's condition and likely prognosis, and in communicating their support needs to funding bodies. One parent in our study said

> *"My daughter has very rare disorders with not much known about it. Only a handful of people in the world with it. They say she is not delayed enough for NDIS funding".*

For those with a relatively well-known diagnosis there is significant precedent and documentary evidence available to decision makers to support their likely prognosis and ongoing support needs, and therefore demonstrate eligibility for funding support. For those with a rare diagnosis or no diagnosis this evidence is lacking, and in many cases the conditions are so rare that the long-term prognosis is unknown. As a result, participants reported that obtaining extensive reports and documentation from health professionals to demonstrate their child's support needs was a key facilitator in gaining access to funding, however this was not always successful.

Participants in this study strongly felt that eligibility for funding is easier to demonstrate for well-known conditions, however further research is needed to understand the extent and accuracy of this perceived inequity.

Alongside poor awareness of schemes and difficulty proving eligibility, systemic issues relating to application processes and the knowledge and capacity of those administering the schemes are also barriers to access.

As with previous studies, many participants found the process of applying for and maintaining access to government funding to be challenging and burdensome [29]. The administrative burden to access government support included repetitive questioning, long and complicated forms, needing to obtain supporting documents, and having to make multiple application attempts due to being considered

ineligible or application forms going missing once submitted. These complex processes contribute to the psychological stress and isolation for parents and caregivers, who are often already experiencing high levels of stress [16, 30]. A lack of services exists to provide support for those living with and caring for individuals with a rare condition which can cause affected individuals to feel overwhelmed and unsupported [8, 31]. In this study, as in previous studies, some families lack the capacity or desire to proceed with applications due to these complicated and time consuming processes, and thus may miss out on funding support to which they are entitled [15].

The lack of knowledge and understanding of rare diseases from staff administering funding schemes and determining eligibility was identified as a potential barrier by parents and guardians in the study. Open communication and generating trust between staff and individuals with a rare condition and their family are necessary to facilitate mutual understanding [25, 31]. Through applying a person-centered care approach where parents caring for individuals with a rare condition are respected will help to avoid a systematic tick box approach which has been heavily felt by many in our study.

### Impacts

Barriers to funding results in significant impacts for both children and families, including delayed or lack of access to needed supports, or forcing families to utilise funds out of their own pocket to compensate for a lack of funding, leading to potential financial stress [15].

Many households identified respite and caregiving support, and lack of access to support for social and community participation as the greatest unmet needs. These unmet needs had subsequent impacts on child participation in activities and household financial stress, caregiver wellbeing and family relationships, which is consistent with previous studies [9, 31]. Enabling access to sufficient funding to meet these support needs is necessary to ensure the health and social wellbeing of individuals living with a rare disease and their families [15]. The impact on parent or caregiver mental health is significant and also supported by previous studies [20], with greater attention needed on providing adequate support to ensure care for the caregivers who often are experiencing significant emotional, social and financial stress[20, 31, 32].

Insufficient funding has significant impacts, but delayed access can also have profoundly negative impacts. As rare diseases can be life-limiting, any delays with applications and wait times to receive funding can be catastrophic [12, 15]. This was highlighted by one of the participants in our study whose child passed away whilst waiting for the outcome of their government funding application and did not ever receive the support they needed.

The barriers identified in this study are consistent with priorities for action identified in the National Strategic Action Plan for Rare Diseases to increase awareness of rare conditions and improve access to support for people living with rare conditions and their families [18]. The study findings, and solutions identified below, also align with those of the recent independent Review into the National Disability Insurance Scheme and subsequent recommendations, including those to: create fairer, more consistent pathways for participants; implement supports for navigating systems; create a continuum of support for children and families; and embed a highly skilled, person-centred, disability aware culture across agencies.[33]

Overcoming the barriers identified in this study is vital to ensure children with undiagnosed and rare genetic conditions have timely and appropriate access to funding which enables them to access the supports they, and their families, need to live their best possible lives.

Some solutions to these barriers include:

- Ensure clear and accessible information that increases parent's/caregiver's awareness of what funding and support is available to them from when a child's is first identified as not meeting their milestones or has health concerns, even in the absence of a diagnosis.

- Information about funded supports, health and disability information including support groups, is promoted to frontline health professionals (e.g. GPs, paediatricians, Maternal & Child Health Nurses, allied health professionals) who may be first point of contact.
- Implement support for families to navigate funding application processes, including for appealing decisions. This may include developing peer support networks for families navigating application processes, parent educations sessions specific to applying for funding for children with rare and undiagnosed genetic conditions, or an online resource repository to share templates of how to complete application forms.
- Build capacity of those agencies and workers responsible for assessing funding applications and determining eligibility for funding support to better understand the need and complexities for children with undiagnosed and rare genetic conditions. This could include access to a Rare Disease System navigator or focal point, who could support undiagnosed and rare disease families as well as Government Service Agencies and their staff.
- Funding eligibility should be determined on function and support needs, not diagnosis. Remove the defined disability lists, which guarantee automatic eligibility and acceptance into the NDIS.
- Provision for interim funding while waiting for a diagnosis to reduce the uncertainty for these families, who are already experiencing the uncertainty of a diagnosis that is either unknown or rare with little information on their child's prognosis.

## Conclusion

This study was the first known survey which focused on the experience of families caring for children with an undiagnosed or rare genetic condition accessing government financial support across Australia. Our findings found most SWAN households have experienced challenges with accessing government funded support and suggests this results in significant unmet support needs for both children and their families. The study highlights that although there many different rare genetic conditions, many families experience similar challenges in accessing funding including lack of knowledge of existing schemes, difficulty demonstrating eligibility in the face of a rare diagnosis or no diagnosis, complex application processes, and lack of understanding of rare genetic conditions by funding bodies.

The findings from this study and proposed solutions could be used by decision-makers and organisations supporting children with undiagnosed and rare conditions to improve equitable access to government funding programs for these children and their families.

As this study was undertaken primarily with SWAN members, further research is needed to understand the experiences of those who are not connected to support and advocacy groups, who are likely to face greater challenges.

### Limitations & Future research

A key strength of our study was the contribution of information to an under-researched topic and improved understanding of families caring for children with an undiagnosed or rare condition access to government funded support. Our study cohort were predominately SWAN members that were sampled from SWAN Australia, an advocacy and support organisation and lacked a comparator population.

Our study findings are therefore skewed towards those who have access to advocacy or peer support organisations like SWAN Australia. The findings are not likely to represent individuals with a rare genetic condition and their families who have not been linked to advocacy organisations such as SWAN. These households may be at a greater disadvantage compared to our study sample, therefore further study is needed to examine their experiences, including families from culturally and linguistically diverse groups that were not captured by this study. A disproportionate number of respondents were females (96.1%), as also seen in previous studies [5, 34]. Findings are therefore more likely to represent female parents, guardians or carers, and those who are willing to participate in research.

In hindsight, we recognised our data should have been categorised as children aged between 16 to 18 years rather than children 13 – 17 years, to allow for more accurate analysis of those accessing DSP and ESS which are eligible for those 16 years and older [2]. This may have impacted our analysis as it could have included some individuals who were not yet eligible for DSP and ESS (i.e., under the age of 16 years)[2].

Further research that aims at including the experiences of individuals with common disabilities or individuals with a rare disease who have not been linked to advocacy organisations would be insightful to understand whether any similarities or differences which may exist in comparison to our study sample.

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**Contact: Fleur Smith smith.f@unimelb.edu.au**